Genetic Diagnostic Area
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Disease	Gen	Description
46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	*SRD5A2*	Sequence analysis of the encoding exons
Abetalipoprotein B deficiency (ABL)	*MTTP*	Sequence analysis of the encoding exons
Aceruloplasminemia	*CP-001*	Sequence analysis of the encoding exons
Achrondroplasia	*FGFR3*	Analysis of G1138A, G1138C, G375C mutations
Achrondroplasia	*FGFR3*	Sequence analysis of the encoding exons
Adenomatous polyposis	*APC*	MLPA
Adenomatous polyposis	*APC*	Sequence analysis of the encoding exons
Adrenal Hypoplasia congenital, 21-hydroxylase deficiency	*CYP21A2*	Sequence analysis of the encoding exons
Adrenal Hypoplasia congenital, 21-hydroxylase deficiency	*CYP21A2*	Sequence analysis of the encoding exons and MLPA
Adrenoleukodystrophy (X- linked)	*ABCD1*	Sequence analysis of the encoding exons
Dominant and recessive polycystic kidneys disease	*PKD1, PKD2, PKHD1*	Sequence analysis of the encoding exons (NGS)
Age-Related macular degeneration (AMD)	*CFH, CFB, C3, ARMS2*	Genetic factors involved in AMD
Aicardi-Goutières syndrome	*TREX1*	Sequence analysis of the encoding exon (1 exon)
Alkaptonuria	*HGD*	Sequence analysis of the encoding exons
Alzheimer	*ApoE*	Sequence analysis of the exon 4
Alzheimer	*APP*	Sequence analysis of the exon 16 and 17
Alzheimer	*PSEN1*	Sequence analysis of the encoding exons
Alzheimer	*PSEN2*	Sequence analysis of the encoding exons
Alzheimer, - Panel	*PSEN1, PSEN2, ApoE, APP*	Sequence analysis of several genes
Alzheimer, Dementia	*CLU*	Sequence analysis of the encoding exons
Alzheimer, Parkinson	*SNCA*	Sequence analysis of the encoding exons
Amyotrophic lateral sclerosis	*PFN1*	Sequence analysis of the encoding exons
Amyotrophic lateral sclerosis type 1	*SOD1*	Sequence analysis of the encoding exons
Analysis of one exon (amniotic fluid sample)	-	Sequence analysis of the encoding exons

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Analysis of one exon (blood or buccal bruses sample)	-	Sequence analysis of one exons
Androgen receptor	*AR*	Sequence analysis of the encoding exons
Angelman syndrome		Sequence analysis of the encoding exons
Angiotensin converting enzyme (ACE)	*ACE*	Analysis of 106180 I/D ACE polymorphism
Aniridia	*PAX6*	Sequence analysis of the encoding exons
Anophtalmia / microphthalmia	*SOX2*	Sequence analysis of the encoding exons
Anophthalmia / Microphtalmia, Mattew-Wood	*STRA6*	Sequence analysis of the encoding exons
Apolipoproteine A-V (APOA5)	*APOA5*	Analysis of 1131T>C mutation
Ataxia telangiectasia	*ATM*	Sequence analysis of the encoding exons and MLPA
Ataxia, Friedreich like, with isolated vitamin E deficiency	*ATTP*	Sequence analysis of the encoding exons
Ataxia-Oculomotor Apraxia, Type 1	*APTX*	Sequence analysis of the encoding exons
Ataxia-Oculomotor Apraxia, Type 1	*APTX*	Sequence analysis of the encoding exons and MLPA
Ataxia-Oculomotor Apraxia, Type 2	*SETX*	Sequence analysis of the encoding exons
Ataxia-Oculomotor Apraxia, Type 2	*SETX*	Sequence analysis of the encoding exons and MLPA
Atrophy muscular espinal	*SMA1, SMA2*	MLPA
Atrophy muscular espinal	*SMA1, SMA2*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS)	*THBD*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS)	*CFB*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS)	*CFI*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS)	*MCP*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS)	*CFH*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS)	*C3*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS) - Panel 1	*CFH, CFI, MCP*	Sequence analysis of the encoding exons
Atypical hemolytic-uremic syndrome (aHUS) - Panel 2	*C3, CFB, THBD*	Sequence analysis of the encoding exons

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Disease	Gen	Description
Atypical hemolytic-uremic syndrome (aHUS) - Panel 3	*CFH, CFI, MCP, CFB, C3, THBD*	Sequence analysis of the encoding exons
Autoimmnue Lymphoproliferative Syndrome	*FASLG*	Sequence analysis of the enconding exons
Autoimmnue Lymphoproliferative Syndrome	*NRAS*	Sequence analysis of the enconding exons
Autoimmnue Lymphoproliferative Syndrome	*FAS*	Sequence analysis of the enconding exons
Autoimmnue Lymphoproliferative Syndrome	*CASP10*	Sequence analysis of the enconding exons
Autoimmnue Lymphoproliferative Syndrome	*CASP8*	Sequence analysis of the enconding exons
Paroxysmal dyskinesia	*PRRT2*	Sequence analysis of the encoding exons
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	*SACS, AFG3L2, MTPAP*	Sequence analysis of the encoding exons (NGS)
Bartter syndrome type 1	*SLC12A1*	Sequence analysis of the encoding exons
Bartter syndrome type 2	*KCNJ1*	Sequence analysis of the encoding exons
Bartter syndrome type 3	*CLCNKB*	Sequence analysis of the encoding exons
Bartter syndrome type 4	*BSND*	Sequence analysis of the encoding exons
Beals	*FBN2*	Sequence analysis of the enconding exons
Becker muscular dystrophy	*CLCN1*	Sequence analysis of the encoding exons
Benign familial infantile epilepsy	*SCN2A*	Sequence analysis of the encoding exons
Bethlem myopathy	*COL6A1, COL6A2, COL6A3*	Sequence analysis of the encoding exons
Biotin-responsive basal ganglia disease	*SLC19A3*	Sequence analysis of the encoding exons
Borjeson-Forssman-Lehmann , síndrome de	*PHF6*	Sequence analysis of the encoding exons
Breast and ovarian cancer	*BRCA1, BRCA2*	BRCA1 or BRCA2 MLPA
Breast and ovarian cancer (NGS)	*BRCA1, BRCA2*	Sequence analysis of the encoding exons (NGS)
Brugada, Dravet, GEFS+ syndrome	*SCN1B*	Sequence analysis of the encoding exons
B-thalassemia	*HBB*	Sequence analysis of the encoding exons
Cadasil	*NOTCH3*	Sequence analysis of the encoding exons 2-24
Campomelic dysplasia	*SOX9*	Sequence analysis of the encoding exons
Christianson syndrome	*SLC9A6*	Sequence analysis of the encoding exons

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Disease	Gen	Description
Cone Rod-Dystrophy	*CRX*	Sequence analysis of the encoding exons
Cone Rod-Dystrophy	*RPGRIP1*	Sequence analysis of the encoding exons
Confirmation of one mutation in parental samples (2 samples)	-	Confirmation of detected mutations
Congenital cataract, X-linked	*NHS*	Sequence analysis of the encoding exons
Congenital Lactase intolerance	*LCT*	Sequence analysis of the encoding exons
Congenital myopathy	*ACTA1*	Sequence analysis of the encoding exons
Cornelia de Lange syndrome	*NIPBL*	MLPA
Cornelia de Lange syndrome	*SMC1A*	Sequence analysis of the enconding exons
Cornelia de Lange syndrome	*SMC3*	Sequence analysis of the enconding exons
Cornelia de Lange syndrome	*NIPBL*	Sequence analysis of the enconding exons
Cowden, síndrome de	*PTEN*	Sequence analysis of the encoding exons and MLPA
Creutzfeld-Jacob, Gerstmann-Straussler-Scheinker syndrome, fatal familial insomnia	*PRNP (prion protein)*	Sequence analysis of the encoding exons
Cystic fibrosis	*CFTR*	Analysis of 34 mutations
Cystic fibrosis	*CFTR*	Sequence analysis of the encoding exons
Dentatorubral-pallidoluysian atrophy	*MAGI2*	Sequence analysis of the encoding exons
Detection of hexanucleotide C9ORF72 in Frontotemporal dementia	*C9ORF72*	Quantification of repeat lenght
Dystonia	*DYT1*	Sequence analysis of the encoding exons
Dravet (SMEI) syndrome	*PCDH19*	Sequence analysis of the encoding exons
Dravet (SMEI) syndrome	*SCN1A*	Sequence analysis of the encoding exons
Dravet (SMEI) syndrome	*SCN1A*	Sequence analysis of the encoding exons and MLPA confirmation of parental mutation are incluided if it will be indicated)
Dravet (SMEI) syndrome	*SCN1A, GABRG2*	Sequence analysis of the encoding exons and MLPA for SCN1A
Dravet (SMEI) , GEFS+ syndrome	*GABRG2*	Sequence analysis of the encoding exons
Duchenne/Becker muscular dystrophy	*DMD*	MLPA
Duchenne/Becker muscular dystrophy	*DMD*	Sequence analysis of the encoding exons (NGS)

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Disease	Gen	Description
Ehlers-Danlos syndrome, arthrochalasic type (type 7)	*COL1A1*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, arthrochalasic type (type 7)	*COL1A2*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, classic type	*COL5A1, COL5A2*	Sequence analysis of the enconding exons (NGS)
Ehlers-Danlos syndrome, dermatosparaxis type (tipe 7C)	*ADAMTS2*	Analysis of the mutation p.Q225X
Ehlers-Danlos syndrome, dermatosparaxis type (tipe 7C)	*ADAMTS2*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, hypermobile type (type 3)	*TNXB*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, kyphoscoliotic type (type 6)	*PLOD1*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, progeroid type	*B4GALT7*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, spondylocheiro dysplastic type	*SLC39A13*	Sequence analysis of the enconding exons
Ehlers-Danlos syndrome, vascular type (type 4)	*COL3A1*	Sequence analysis of the enconding exons
Episodic ataxia (EA1)	*KCNA1*	Sequence analysis of the encoding exons
Episodic ataxia (EA1)	*KCNA1*	Sequence analysis of the encoding exons and MLPA
Episodic ataxia (EA2)	*CACNA1A*	Sequence analysis of the encoding exons
Episodic ataxia (EA2)	*CACNA1A*	Sequence analysis of the encoding exons and MLPA
Episodic ataxia (EA5)	*CACNB4*	Sequence analysis of the encoding exons
Episodic ataxia (EA6)	*SLC1A3*	Sequence analysis of the encoding exons
Fabry	*GLA*	Galactosidase a biochemical levels
Fabry	*GLA*	Sequence analysis of the encoding exons
Factor V Leiden	*FACTOR V*	Analysis of R506Q mutation
Familial adenomatous polyposis	*MYH*	Analysis of Y165C, G382C mutations
Familial adenomatous polyposis	*MYH*	Sequence analysis of the encoding exons
Familial hemiplegic migraine	*CACNA1A, ATP1A2, SCN1A*	Sequence analysis of the encoding exons

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Familial hypercholesterolemia Type 3	*PCSK9*	Sequence analysis of the encoding exons
Fragile X	*FMR1*	Microsatellite analysis
Friedreich ataxia	*FRDA (X25)*	Quantification of repeat lenght
Friedreich ataxia	*FRDA (X25)*	Sequence analysis of the encoding exons
Fronto-temporal dementia	*VCP*	Sequence analysis of the encoding exons 3, 5, 6, 7, 8
Fronto-temporal dementia	*TARDBP*	Sequence analysis of the encoding exons
Fronto-temporal dementia	*PGRN*	Sequence analysis of the encoding exons
Fronto-temporal dementia	*MAPT*	Sequence analysis of the encoding exons
Fronto-temporal dementia - Panel	*MAPT, PGRN, FUS, VCP, TARDBP*	Sequence analysis of several genes
Fronto-temporal dementia, Amyotrophic lateral sclerosis	*FUS*	Sequence analysis of the encoding exons 3, 5, 6, 13, 14
Fructosemie	*ALDB*	Analysis of A149P, A174D, N334K mutations
Fructosemie	*ALDB*	Sequence analysis of the encoding exons
Galactosidase, beta 1	*GLB1*	Sequence analysis of the encoding exons
Gender determination	*SRY*	SRY detection
Hedgehog (Holoprosencephaly type 3)	*SHH*	Sequence analysis of the enconding exons
Hemochromatosis	*HFE*	Analysis of C282Y, H63D, S65C mutations
Hemochromatosis	*HFE*	Sequence analysis of the encoding exons
Hemophagocytic lymphohistiocytosis	*STX11*	Sequence analysis of the encoding exons
Hemophagocytic lymphohistiocytosis	*PRF1*	Sequence analysis of the encoding exons
Hemophagocytic lymphohistiocytosis	*STXBP2*	Sequence analysis of the encoding exons
Hemophagocytic lymphohistiocytosis	*UNC13D*	Sequence analysis of the encoding exons
Hemophagocytic lymphohistiocytosis	*UNC13D, STX11, STXBP2*	Sequence analysis of the encoding exons
Hereditary neuropathy with liability to pressure palsies (NHPP)	*PMP22*	Sequence analysis of the encoding exons
Hereditary nonpolyposis colorectal cancer	*MLH1, MSH2, MSH6*	MLH1 or MSH2 MLPA
Hereditary nonpolyposis colorectal cancer	*MLH1, MSH2, MSH6*	Microsatillite instability

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Hereditary nonpolyposis colorectal cancer	*MLH1, MSH2, MSH6*	Analysis of MLH1 or MSH2 or MSH6 mutations
Hereditary nonpolyposis colorectal cancer	*MLH1, MSH2, MSH6*	sequence analysis for several genes
Holoprosencephaly	*SIX3*	Sequence analysis of the enconding exons
Holoprosencephaly	*SHH, ZIC2, SIX3, TGIF1*	MLPA
Holoprosencephaly	*ZIC2*	Sequence analysis of the enconding exons
Holoprosencephaly	*TGIF1*	Sequence analysis of the enconding exons
Huntington disease	*HTT*	Quantification of repeat lenght
Hypertrophic cardiomyopathy - Panel NGS 1	*MYH7, MYBPC3, TNNT2, TNNI3, TPM1, TNNC, MYL2, MYL3, ACTC1*	Sequence analysis of several genes (NGS)
Hypertrophic cardiomyopathy - Panel NGS 2	*MYH7, MYBPC3, TNNT2, TNNI3, TPM1, TNNC, MYL2, MYL3, ACTC1, CAV3, PRKAG2, LAMP2, GLA, TTR*	Sequence analysis of several genes (NGS)
Imperfect osteogenesis	*COL1A1*	Sequence analysis of the encoding exons
Imperfect osteogenesis	*COL1A2*	Sequence analysis of the encoding exons
Increased in melanoma	*miR211*	Sequence analysis of the encoding exons
Joubert syndrome	*NPHP1*	Sequence analysis of the encoding exons
Joubert syndrome	*AHI1*	Sequence analysis of the encoding exons
Joubert syndrome, Leber syndrome	*CEP290*	Sequence analysis of the encoding exons
Kabuki syndrome	*MLL2*	Sequence analysis of the encoding exons
L-DOPA responsive dystonia	*DYT5*	Sequence analysis of the encoding exons
Lactose intolerance (adults)	*LCT*	Analysis of 3910T>C and 22018G>A mutations
Larsen, síndrome de	*FLNB*	Sequence analysis of the encoding exons
Leber Congenital Amaurosis	*LRAT*	Sequence analysis of the encoding exons
Leber Congenital Amaurosis	*AIPL1*	Sequence analysis of the encoding exons
Leber Congenital Amaurosis	*RPE65*	Sequence analysis of the encoding exons

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Leber Congenital Amaurosis	*LCA5*	Sequence analysis of the encoding exons
Leber Congenital Amaurosis	*RDH12*	Sequence analysis of the encoding exons
Leber Congenital Amaurosis	*GUCY2D*	Sequence analysis of the encoding exons
Lens and retinal development	*miR204*	Sequence analysis of theno encoding exon
Leukoencephalopathy with vanishing white matter	*EIF2B5*	Sequence analysis of the encoding exons
Long QT syndrome, Andersen-Tawil syndrome	*KCNJ2*	Sequence analysis of the enconding exons
Long QT syndrome, Jervell and Lange-Nielsen syndrome	*KCNQ1, KCNE1*	Sequence analysis of the enconding exons
Long QT syndrome, Romano-Ward syndrome	*KCNE1, KCNE2*	Sequence analysis of the enconding exons
Long QT syndrome, Romano-Ward syndrome	*KCNQ1, KCNH2, SCN5A*	Sequence analysis of the enconding exons
Long QT syndrome type 2	*KCNH2*	Sequence analysis of the encoding exons
Macular distrophy, Retinitis pigmentosa	*PDE6A*	Sequence analysis of the encoding exons
Marfan syndrome	*TGFB2*	Sequence analysis of the encoding exons
Marfan I syndrome	*FBN1*	Partial sequence analysis of the encoding exons (65% of detection)
Marfan I+II syndrome	*FBN1*	Sequence analysis of the encoding exons
Marfan II syndrome	*FBN1*	Partial sequence analysis of the encoding exons (35% of detection)
Marfan syndrome - Panel	*FBN1, FBN2, TGFBR2*	Sequence analysis of the encoding exons (NGS)
Marinesco-Sjögren syndrome	*SIL1*	Sequence analysis of the encoding exons
Methylenetetrahydrofolate-reductase	*MTHFR*	Analysis of 677C>T and 1298A>C mutations
Microdeletion of Y chromosome	*AZF*	AZFa, AZFb, AZFc deletions
Microphthalmia, nonophthalmos	*MFRP*	Sequence analysis of the encoding exons
Myopathy, myocardiopathy	*MYH7*	Sequence analysis of the encoding exons
Mitochondrial DNA (DNA purified)	-	On request
Mitochondrial DNA (sample from blood or biopsy)	-	DNA isolation and analysis of mitochondrial genome
Mitochondrial DNA (sample from blood or biopsy)	-	DNA isolation, analysis of mitochondrial genome and haplogroup

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Morquio syndrome, Mucopolysaccharidosis type IVa	*GALNS*	Sequence analysis of the encoding exons
Multiminicore myopathy, centronuclear myopathy (NGS)	*RYR1*	Sequence analysis of the encoding exons
Myeloproliferative disease	*MPL*	Analysis of W515L mutation
Myoclonic astatic epilepsy, childhood absence epilepsy. GLUT1 deficiency syndrome	*SLC2A1*	Sequence analysis of the encoding exons
Myophosphorylase deficiency (McArdle disease,GSD5)	*PYGM*	Analysis of p.R50X mutation
Myophosphorylase deficiency (McArdle disease,GSD5)	*PYGM*	Sequence analysis of the encoding exons
Myotonia congenita	*CLCN1*	Sequence analysis of the enconding exons
Myotubular myopathy	*MTM1*	Sequence analysis of the enconding exons
Neurofibromatosis I	*NF1*	MLPA
Neurofibromatosis I	*NF1*	Sequence analysis of the enconding exons
Niemann-Pick	*NPC1, NPC2*	Sequence analysis of the encoding exons
Night blindness	*PDE6B*	Sequence analysis of the encoding exons
Nodular heterotopia	*FLNA*	Sequence analysis of the encoding exons
Non-syndromic deafness	*GJB2, GJB6, OTOF, ARNr12s, USH2A**	Screening for prevalent mutations (12) in GJB2, GJB6, OTOF genes, and in the gene that encoded ARNr 12s
Noonan syndrome	*PTPN11*	Sequence analysis of the encoding exons
Oculodigitoesophagoduodenal Syndrome	*MYCN*	Sequence analysis of the encoding exons and MLPA
Oguchi Disease	*SAG*	Sequence analysis of the encoding exons
Ohtahara syndrome	*STXBP1*	Sequence analysis of the encoding exons
Ohtahara syndrome	*STXBP1, ARX*	Sequence analysis of the encoding exons
Optic Atrophy 1	*OPA1*	Sequence analysis of the encoding exons
P450 cytochrome	*CYP2C19*	Sequence analysis of the encoding exons
Paramyotonia congenita (Feingold)	*SCN4A*	Sequence analysis of the encoding exons
Parkinson	*PARK8 / LRRK2 (ex. 31, 34, 35, 41,*	Sequence analysis of the encoding exons

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Perry syndrome (mutations in exon 2)	*DCTN1*	Sequence analysis of the encoding exons
Perry syndrome (all codifying exon except exon 2)	*DCTN1*	Sequence analysis of the encoding exons
Plasminogen activator inhibitor-1 (PAI-1)	*PAI1*	Analysis of 675 4G/5G and 844A>G polymorphism
Plasminogen activator inhibitor type 1 deficiency	*PAI1*	Sequence analysis of the encoding exons
Postnatal array (180k)	*Array*	CGH array
Postnatal array (400k)	*Array*	CGH array
Postnatal array (60k)	*Array*	CGH array
Prader-Willi / Angelman	*PW / AS*	MLPA
Produced by tumor cell line and by macrophages	*CCL7 (MCP3)*	Sequence analysis of the encoding exons
Protombine (Factor II)	*FACTOR II*	Analysis of 20210G>A mutation
Pseudoxanthoma elasticum	*ABCC6*	Sequence analysis of the encoding exons and MLPA
Pulmonary alveolar microlithiasis	*SLC34A2*	Sequence analysis of the encoding exons
Rendu-Osler-Weber (hemorrhagic telangiectasia, HHT)	*ENG, ALK1*	Sequence analysis of the encoding exons and MLPA
Retinitis pigmentaria	*RGR*	Sequence analysis of the encoding exons
Retinitis pigmentaria	*CNGB1*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP10)	*IMPDH1*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP12)	*CRB1*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP14)	*TULP1*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP2)	*RP2*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP26)	*CERKL*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP3)	*RPGR*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP33)	*MERTK*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP37)	*NR2E3*	Sequence analysis of the encoding exons

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Retinitis pigmentaria (RP4)	*Rho*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP41, RP49)	*CNGA1*	Sequence analysis of the encoding exons
Retinitis pigmentaria (RP7)	*RDS (PRPH2)*	Sequence analysis of the encoding exons
Retinitis prigmentaria	*RLBP1*	Sequence analysis of the encoding exons
Rett syndrome, atypical	*CDKL5*	Sequence analysis of the encoding exons
Rett syndrome, congenital variant	*FOXG1*	Sequence analysis of the encoding exons
Rett syndrome, congenital variant	*FOXG1*	Sequence analysis of the encoding exons and MLPA
Rett syndrome, typical	*MECP2*	Sequence analysis of the encoding exons and MLPA
Rheumatoid arthritis and inflamatory response againist tumor	*CCR2 (CD192)*	Sequence analysis of the encoding exons
Risk factor for early pregnancy termination	-	Analysis of mutation in Prothrombin (G20210A), MTHFR (C677), and Leiden's Factor V (R506)
Spastic paraplegia recessive type 1	*L1CAM*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 10	*KIF5A*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 11	*KIAA1840*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 15	*ZFYVE26*	Sequence analysis of the encoding exons
Spastic paraplegia type 17 (Silver syndrome) (mutations in exon 3)	*BSCL2*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 2	*PLP1*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 3	*SPG3A*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 4	*SPAST*	Sequence analysis of the encoding exons
Spastic paraplegia recessive type 7	*SPG7*	Sequence analysis of the encoding exons
Spinocerebelar ataxia (PANEL SCA1, SCA2, SCA3, SCA17)	*ATXN1, ATXN2, ATXN3, TBP*	Quantification of repeat lenght
Spinocerebelar ataxia (PANEL SCA1, SCA2, SCA3, SCA6, SCA7)	*ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7*	Quantification of repeat lenght
Spinocerebelar ataxia 1 (SCA1)	*ATXN1*	Quantification of repeat lenght
Spinocerebelar ataxia 10 (SCA10)	*ATXN10*	Quantification of repeat lenght

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Spinocerebelar ataxia 12 (SCA12)	*PPP2R2B*	Quantification of repeat lenght
Spinocerebelar ataxia 15 (SCA15)	*ITPR1*	Sequence analysis of the encoding exons
Spinocerebelar ataxia 17 (SCA17)	*TBP*	Quantification of repeat lenght
Spinocerebelar ataxia 18 (SCA18)	*IFRD1*	Sequence analysis of the encoding exons - PROPOSED gene
Spinocerebelar ataxia 2 (SCA2)	*ATXN2*	Quantification of repeat lenght
Spinocerebelar ataxia 27 (SCA27)	*FGF14*	Sequence analysis of the encoding exons
Spinocerebelar ataxia 3 (SCA3), Machado-Joseph disease	*ATXN3*	Quantification of repeat lenght
Spinocerebelar ataxia 5 (SCA5)	*SPTBN2*	Sequence analysis of the exon 7
Spinocerebelar ataxia 5 (SCA5)	*SPTBN2*	Sequence analysis of the encoding exons
Spinocerebelar ataxia 6 (SCA6)	*CACNA1A*	Quantification of repeat lenght
Spinocerebelar ataxia 7 (SCA7)	*ATXN7*	Quantification of repeat lenght
Spinocerebelar ataxia 8 (SCA8)	*ATXN8OS*	Quantification of repeat lenght
Thrombotic thrombocytopenic purpura by reduction in ADAMTS13	*ADAMTS13*	Sequence analysis of the encoding exons
Tissue injury, infection and inflammation	*CCL2 (MCP1)*	Sequence analysis of the encoding exons
Tuberous sclerosis	*TSC1*	MLPA
Tuberous sclerosis	*TSC2*	MLPA
Tuberous sclerosis	*TSC1*	Sequence analysis of the encoding exons
Tuberous sclerosis	*TSC2*	Sequence analysis of the encoding exons
Tumor protein p53	*TP53*	Sequence analysis of the encoding exons
Vascular dementia - Panel	*NOTCH3, TREX1*	Sequence analysis of several genes
Vitreoretinopathy	*LRP5*	Sequence analysis of the encoding exons
Von Hippen Lindau, síndrome de	*VHL*	Sequence analysis of the encoding exons
West syndrome	*ARX*	Sequence analysis of the encoding exons
West syndrome	*ARX, CDKL5*	Sequence analysis of the encoding exons
wolfram syndrome	*CISD2*	Sequence analysis of the encoding exons

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Disease	Gen	Description
wolfram syndrome	*WFS1*	Sequence analysis of the encoding exons
wolfram syndrome	*WFS1, CISD2*	Sequence analysis of the encoding exons

If you have any other diagnostic genetic test that is not in this catalog, please contact us without any obligation.
Prices are valid from April 1, 2013.
Approximate response time 30 days (Consult for each diagnostic).
Required Sample: 10-15 cc blood in EDTA tube (Consult for other samples).